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## The Truth About Down Syndrome

### **SUMMARY KEYWORDS**

down syndrome, children, parents, support, test, mothers, gut motility, families, hypothyroidism, extra chromosome, bit, chromosome, diagnosis, common, accuracy, translocation, therapy, medical, individuals, gastrointestinal issues

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Welcome to our session on Down syndrome and pregnancy. With me is Dr. Erica Peirson, who is the medical director of the Peirson center for children, which focuses on children with special needs. Dr. Peirson, thanks so much for joining us today.

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Yeah, thanks for inviting me. This is wonderful.

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If you could tell us a little bit about your professional background and your personal interest in the topic.

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Yeah, thanks. So, so I'm a naturopathic physician and my personal interest in this topic began with the birth of my own son, who does have mosaic Down syndrome. And so naturally, I, you know, immediately wanted to help him and dove into the research and in what's available to help children with Down syndrome. There's a lot of research to support the work that I do today. And so I've studied, you know, extensively, I think is an understatement. Everything that's available to help our children and learned kind of my own things that I hope to share with with you and your audience today.



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That's great. So let's just begin with if you could tell us a little bit about what is Down syndrome and the different forms of Down syndrome.

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Yeah, so Down syndrome is results from an extra copy of chromosome 21. It is a very small chromosome as the chromosomes are numbered from, you know, largest to smallest, and we have 23 pairs of chromosomes. So, chromosome 21, and extra copies is Down syndrome. And there are different forms of edits. It's really from a medical perspective, it's it's quite fascinating what, what happens with our body when, when you know, babies are created. So that extra chromosome can either be in isolation on its own, there are just three copies of it. There's also translocation Down syndrome where a piece of that chromosome 21, particularly within what we call the Down syndrome, critical region of chromosome 21. So you don't need the whole chromosome can attach itself to another chromosome. So we get a little piece of it. And then there's a that's only 4% of cases of Down syndrome will have translocation and there can be a genetic component. Maybe a parent carries that translocation, they're not aware of it, perhaps but it is quite rare. And then even more rare, about 1% of cases of Down syndrome have what my son has, which is mosaic Down syndrome. And that's where the the division of that extra chromosome occurs later on within the cell division, not at the initial cell division, and it results in the extra chromosome being in just a certain percentage of the cells of that child. And it's, they're extremely variable.

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Yeah. Well, thanks for explaining that genetics. Can you also explain a little bit about how the genetics affects individuals with Down syndrome?

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Yeah, it's hugely variable, right, which is where we maybe we'll get into later on, you know, the causes that variability in terms of what the term we use is phenotype and phenotype is how a person Present, right? Like I have brown hair, we are of a certain height, right? This is our phenotype, what we look like. And so the phenotype of individuals with Down syndrome, like I said, is hugely variable. You know, largely because of all the other factors that make up who we are. There are other chromosomes, their environment. And so, so to answer your question, yeah, they, you know, they can all appear very differently have different levels of, you know, function, different levels of cognition. So there's a lot of variability there for sure. Yeah.

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Thank you. So one of the factors that can increase the chance that a couple will conceive a baby with down syndrome.



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Yeah, so again, there's a fair amount of research there. I would say if I could narrow it down to maybe just a few things. What we would typically learn in medical school is the age you know, advanced age of a mother, I would say past 35/36. There's definitely a little steep increase in the chance of conceiving a child with an extra chromosome, but we like to look deeper than that, you know, what is it about the aging process that, you know changes the those chances. Some of it is hypothyroidism in others, you know, perhaps even subclinical hypothyroidism has been linked to increasing chances of having a child with Down syndrome. Another factor is methylation, right which is a huge topic unto itself. But genetically within a parent, particularly the mothers right there's a lot of you know, we've we've we carry a big role in this for sure the mothers and our eggs, but having a genetic single nucleotide polymorphism, a change in a gene in a gene called mthfr, which some of your families may be familiar with some not changes the way the body uses foliate right. And if you're carrying an mthfr You know, variation in your gene and you're not able to process full eight properly, that increases the risk of having a child with Down syndrome. And of course, there are many other factors impacting methylation, hypothyroidism impacts methylation, for sure. And then to take it one little bit of a step further. I've also read some research and, you know, given the work that I've done in my understanding of some biochemistry here, glucose metabolism is a big topic and there's some research looking at thiamine deficiency in mothers perhaps contributing to alterations in in that, you know, conception process that can increase the chance of having a child with Down syndrome. Yeah.

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Yeah, so certainly age is a big factor, but nutritional factors are looking at Foley, folic acid, very important as well. And then, um, can you talk a little bit about prenatal testing? How accurate is it? What sort of tests available today.

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Yeah, so today, I believe what it came on the market around 2011. It's called a non invasive prenatal testing and IPT. And they're using free cell believe it's free cell DNA. So it's really fascinating. When we have children, DNA from our children gets into our body, right? And it's travels all throughout our body, we carry it our mothers do throughout our lives, right? It's kind of a beautiful thought, actually. And so what they do is they will take a blood sample from the mother and see if there's any DNA from the fetus, indicating that there are some genetic anomalies right like that we will test for Down syndrome extra chromosome 21. Edwards syndrome, which is an extra copy of chromosome 13 and putao syndrome, extra chromosome 18 and others I believe, and so, so in terms of the accuracy of this testing, because is being offered widely. I believe it was initially designed to be offered to mothers of more advanced age beyond 35 perhaps. But again, my understanding is that is it is offered, you know, to all aged



women. And the accuracy is, I believe, debatable of these tests. There's a lot that can interfere with the accuracy of that DNA floating around that in the mother's blood. You can have, for example, you can have an extra chromosome in the placenta, but it not be in the baby. Right again, as the mother of somebody have a child with mosaic Down syndrome, I have an understanding of how that division can occur in different tissues can have the extra chromosome or not. So just as one example of something that can happen, there are also changes in the mother's cells and DNA that can that are coming from the mother, not from the baby. That can interfere with the test. And so in terms of the accuracy, what's what's often used in terms of the ratio of accuracy is that it's 98 to 99% accurate. And those numbers are coming from the manufacturers of the test. And so I would personally argue that, you know, the trusting the manufacturers of a test who are, you know, perhaps, in who are benefiting from, you know, financially from this test being used. So I don't I choose not to look at those numbers necessarily. And there's other information out there. You know, saying that as much as 50% of the test results can be false positives, meaning that the test is showing you a chip that you have an increased chance of that child having Down syndrome and in fact that child does not have Down syndrome perhaps, and there are reports online have, you know, parents, you know, even terminating a pregnancy when the child didn't even have a You know, chromosome anomaly. So so the the rate of accuracy of the test is debatable and i would i would share some resources here. There's a gentleman Mark Leach. He's one of the experts here. His website is [Downsyndromeprenataltesting.com](http://Downsyndromeprenataltesting.com), and he has some good, you know, well cited scientific information on on the accuracy of the test. So there are tool, but but there I would argue that 99% accuracy is not accurate. Yeah,

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you're saying that the blood tests, it's nice because it's not very invasive. So it's safe test to do, but you may get some false positives. And so if you have a false if you have a positive test, what would you recommend this next step additional testing?

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additional testing? Absolutely. Now, some of that testing can be invasive and amniocentesis, for example would be a potential next test. there is an increased rate of, you know, spontaneous abortion from, from an amniocentesis. So it's an invasive test, you know, they're putting a needle into the amniotic fluid and pulling out cells there. So yeah, so parents should be prepared. I don't think these tests should be used as casually sometimes as they are. And and then the the there is a lack of support. I would argue also that once that diagnosis or that chance is made, and that diagnosis is made, that information that parents are given and the support they're given after that information is shared, the diagnosis potential diagnosis is shared. The support isn't always there. Nor is there accurate information given to parents about



what it really means to have a child with Down syndrome that we may or may not get into today. Yeah.

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Well, let's talk a little bit about that. So how is the news of the diagnosis generally conveyed to parents they hear about a positive test result, but may need conference From additional testing,

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yeah, so we've, so we talked with I talk with families all day, right when I see patients, and I'll just start again. So I talk with families all day. And, you know, when I'm seeing patients and many of them are new families, and I always want to hear their story, it doesn't change how I help them medically, but I just want to hear their story of how their diagnosis was delivered. Because when that diagnosis is delivered, however, whoever does it really sets the tone for parents, right? Many parents maybe even have never heard of Down syndrome. They don't know what it means. And they're looking for that, that expert, to almost tell them how to feel about it, you know, and so when that diagnosis is delivered in a tone of, I am so sorry, I hate to tell you this terrible news, right? It's almost telling the parents how they should feel about it. And it doesn't happen. It's it's you know, it's it's It's it's very big information to give to a parent and it is it does change the trajectory trajectory of that child's life to, you know, to have an extra chromosome. But if you talk with any parent today of a child with Down syndrome, they will tell you that it is not the bleak kind of life and path that they were told that their child would have. In fact, there was a there was research done, I think in the last five years fairly current, where they surveyed several thousand parents. And what they found was an overwhelming majority, I believe 79% or more parents and siblings believed that they had a positive experience based on having a sibling or a child and their outlook on life was more positive because of their child. So this is information that isn't always shared with with families who are just receiving the diagnosis of what it really means to have a child or an adult or a person in your life. With with down syndrome, so, yeah, so that support isn't always there. Yeah.

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Yeah. So if you had a family who had just received the diagnosis of Down syndrome, what would you tell them briefly?

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Yeah, so I would say that, you know, history has has not been been kind to those with down syndrome and and if we look at the historical perspective of what it means to have Down syndrome, a lot of it is based on society's, you know, kind of lack of acceptance and not sort of creating a place at the table for them as it were. And so today, the opportunities that are there for individuals with Down syndrome are so much greater than they were even 20/30 years ago.



And we, those of us parents, who are here today, you know, with our children are so so grateful. I often get emotional here with two parents who have gone before us who have broken those boundaries and those obstacles and you know, and pushed for their children's and In school and in society, and so I would tell new parents who perhaps are receiving a diagnosis, that the opportunities for their children today are a lot different than what children had in the past. So yeah, so that's that's one, you know, from an emotional perspective, I suppose. Yeah.

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Certainly a lot more medical support today a lot more educational and other supports to help raise children. So what is it like to raise a child with Down syndrome? Can you tell us a little bit about that?

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Yeah, so, um, I don't want to sugarcoat it, right? Um, but in many ways, it's like any other parenting of any other child, right? We support our children. We, you know, we encourage them, we keep our expectations as high as possible as you would with any child, right. And so, in many ways, it's very much the same, right? We are included within families. You know, they're loved within their community. And, you know, however right there are doctor's appointments and you know, medical things to keep, keep an eye out for there are certain screenings that we have to be mindful of for our children. But where we run into obstacles, in my experiences is often with, you know, that inclusion in society and acceptance and but within the family unit. You know, in many cases, not every case, it's it's a lot like, you know, any other family,

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right? Sure.

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Sure. So, can you talk a little bit about what are some of the health challenges used to be people with Down syndrome had decreased lifespans, but there's been a lot of medical advances to really improve on that. So what are some of the health challenges that they have?

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Yeah, so first, off the bat, right when a child is born, all children with Down syndrome are screened for heart defects, right. They're very common all children in general screened for heart defects but particularly there's a high you know, radar for that in our children with Down syndrome. Many of them are born with atrial septal defects and vent, you know, ventricular septal defects, etc. So they're screened for that that's a health medical condition to look for. hypothyroidism. Screening should occur regularly for hypothyroidism in children. I think it's often



recommended annually. In my clinic, we look a little bit more closely than that. And I look very closely for symptoms of hypothyroidism, which are often confused with symptoms of Down syndrome. So and then we look in great detail and their thyroid hormone function a little bit more than just a sort of TSH screening. And then and then also other conditions that can impact their overall health or gastrointestinal issues. Right slow gut motility is very common, of course that's linked to hypothyroidism. And then with that slow gut motility come a lot of other kind of sequelae of, you know, malabsorption malabsorption is very common in our population of children. And so gastrointestinal issues. And then just one more thing is, um, sleep apnea. Right? There's a lot of studies looking at sleep apnea in very young children. You know, with down syndrome and screening should occur early before the age of three, it's recommended that a child with Down syndrome have a sleep study to detect even silent sleep apnea, right. And then, of course, the low oxygenation that occurs during sleep apnea can be can impact their, you know, cognition, and overall health. So I think those are some three major issues. There are others that were mindful of mitochondrial dysfunction, right, some more detail here, but a lot of those are rooted in their gastrointestinal issues, I would say, yeah,

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although they may have some medical issues. These are certainly possible to be screened for and possible to be treated? Absolutely.

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Yeah, absolutely.

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Great. And so you've lectured a little bit about diagnostic overshadowing in the past. Could you briefly mention what that is?

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Yeah, so this happens in the autism community as well. And so, and this is important information for new parents that you know, in medical school, right me I was taught all you know, with down syndrome, that it's an extra chromosome happens in older mothers and we're not taught much about it. Other than Yeah, there are some health challenges for those with down syndrome and and i think that because we're medically trained that it's a you know, a bit of a bleak diagnosis and they they're unhealthy and they tend to be unwell. That these that when a child then comes into a medical you know, doctor's office with let's say you know, you know constipation, poor gross, some behavioral or cognitive issues the dial the doctor will will dismiss those symptoms as being caused by the primary diagnosis of Down syndrome or autism or other, you know, health primary diagnosis, when in fact there are other things going on that are being missed that you know can be treated and remedied to help support that child



and optimize their health. So that diagnostic overshadowing it means that the that all of the symptoms and the ill health, let's say, are put into that Down syndrome bucket and kind of dismissed as well. It's just Down syndrome. When in fact, there are a myriad of other things that are being quite honestly missed. And that's where we, you know, in my practice, come in and pick those things up and yeah, work on them. Yeah,

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yeah, so just be as a child with Down has Down syndrome, they may still have other medical challenges that should be looked for assessed and treated. Right. And then um, can you talk a little bit about What are some of the key areas of health that you address in the, in your patients with down syndrome.

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Yeah, I alluded to that a little bit for sure. The

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if I were to pick one, let's say, I would say gastrointestinal issues, you know there in in our children and infants with down syndrome newborns even. There can be some significant nutritional deficiencies in children, and they're often secondary to gastrointestinal issues. I mentioned briefly the, you know, slow gut motility that just because a child is having a bowel movement daily, let's say doesn't mean they don't have slow gut motility we're talking about you know, top to bottom gut motility, microscopic microvilli gut motility, and we can have an increased rate of small intestinal bacterial overgrowth in our children. yeast overgrowth is very common in our children, often secondary to the high rate of antibiotics for our children. So that is a significant area that we help our patients in is optimizing gut motility. And I mean top to bottom gut motility, not just you know, having a daily bowel movement. And then kind of more on an in depth level, you know, we do look at mitochondrial function, mitochondrial being, of course, the powerhouse of the cell. And, you know, creating energy within the body is very important for brain function. So we look at kind of details of mitochondrial function to support our patients. And then, of course, looking very in depth at thyroid hormone labs, nutrient deficiencies, etc. But the gut is really key. Yeah.

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And so then looking at that, looking at their biochemistry, you can then understand how to tailor supplements for each individual based on their differences. There may be some commonalities, but certainly some differences there too.

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Absolutely. Yep. Yeah, absolutely.



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Great. And so it's a very common Sounds like you were alluding to children with Down syndrome having some common vitamin deficiencies and maybe needing extra nutritional support.

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Yes, it's you know, it's it's a little controversial within our community whether to give children with Down syndrome supplements or not as I believe maybe an autism, it is an autism community as well. And all it takes is some simple testing. Yeah, all it takes is some simple testing to be able to pick up you know, their zinc levels are very important to look at. iron deficiency is very common for children. That's the, you know, most well studied, you know, nutrient deficiency globally and linked to lower cognitive levels. He real issues Even so, running some, you know, simple nutritional tests will will help pick these things up for sure.

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Right. So, those are certainly important things to do, and they can help with the outcome of those children and so what supports are available In general to help raise a child with Down syndrome, can you talk a little bit about that? financial support? Government supports educational support?

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Wonderful. Yeah. So there are a lot of opportunities that varies from state to state, right, in terms of kind of governmental support. And, you know, kind of different health insurance plans and and so, yeah, so it varies from state to state. Of course, Medicaid, Medicare, I believe, you know, can be can help support a child with with with down syndrome. So, I'm not as up on the kind of the financial aspect of that, unfortunately.

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But there are, you know, local county

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services or early intervention services that help with you know, physical therapy, feeding therapy, speech therapy, etc. But even beyond that, there's a whole myriad of I would call them alternative therapies but I don't like using that word alternative therapies that the families I work with are seeking out for just a little bit of extra support beyond just you know, conventional physical therapy, different kind of reflex integration and, and different kind of movement therapies for their children. So there's a whole myriad of therapies out there for parents to seek out and what I would, what I would guide those parents to do is to find out what works best for their family and their and they're kind of mindset for their child right? Just because one parent is



choosing a therapy for their child and it's working for them. I don't want parents to ever feel guilty if they can't do this, you know, extremely elaborate, you know, therapy program that one family's choosing, but they can't do. So, to again, that variability right choosing what works best for you and your family. So There are a lot of lot of options out there to help support children with, with their, you know, physical and cognitive development. Certainly.

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Sure. Certainly a lot of government services you mentioned through Medicaid available for children and their families, and then also educational supports in the school district as well. If they need extra educational support, yes. All the medical issues. Sure. So overall, a lot of things to help families if they do have children, that child with Down syndrome, they shouldn't feel overwhelmed by it. today. There's just so much better knowledge about it. So much better medical care, so much better educational supports for those individuals.

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Absolutely. Yeah, absolutely. And yeah, I neglected to mention schools. There are, you know, wonderful support services. There are challenges there as well. Many of us are really kind of pushing for more and more for our children. But those services are there for sure that when If a parent were to receive a diagnosis, they are absolutely not alone. In fact, I would argue that all they have to do is go online. And there is a global, I would say support system. You know, on the internet today are hundreds of thousands of parents, I would literally say that many parents with children with Down syndrome who are looking to support one another Even so, yeah, huge support system for sure.

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That's great. And so what do you see overall for the future of people with Down syndrome?

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Yeah, you know, I see positive things for sure. You know, and I hope that we continue to see individuals with Down syndrome in our society. They be given the, the lessons that we learn from them and the and how they do you know, what they offer our community and society in terms of what they teach us about acceptance of others and how to love one another. It's really you know, quite a bit that we gain from them. So one I hope that we continue to see individuals with Down syndrome in our society but and so given that what what we see, you know, online, you know, in TV shows even that individuals with Down syndrome are capable of and living longer. I just see more and more positive things. And, and, you know, seeing what my fellow mothers out there are doing to advance that for children just gives me great hope and encouragement for for those, you know, who are, you know, coming after us even for sure.



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That's sort of the my perception is that, um, it may be a little bit of that extra challenge to parent a child with Down syndrome. But those individuals and their families can experience a lot of joy as well and can really have a wonderful life for both the child and for the parents and in many ways they can be a blessing.

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Absolutely. Yeah,

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I couldn't agree more. Yeah.

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Great. Well, thank you very much, Dr. Peirson really appreciate your sharing your knowledge with us. Is there anything else you'd like to add? Or is that we pretty much covered everything?

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No, I think I think those are I think those are the highlights. those are those are big messages that I wanted to share. Yeah. And to. Yeah, and to the Down syndrome does not have to be a, you know, a negative thing for their child or their family. And for the most part, it is positive. Yeah.

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Great. Well, thank you so much for sharing all your information with us. Thank you.

